DNA sequence

Genetic testing, also known as DNA testing, is used to identify changes in DNA sequence or chromosome structure.

A nucleic acid sequence is a succession of letters that indicate the order of nucleotides within a DNA (using GACT) or RNA (GACU) molecule. By convention, sequences are usually presented from the 5' end to the 3' end. For DNA, the sense strand is used. Because nucleic acids are normally linear (unbranched) polymers, specifying the sequence is equivalent to defining the covalent structure of the entire molecule. For this reason, the nucleic acid sequence is also termed the primary structure.

The sequence has capacity to represent information. Biological deoxyribonucleic acid represents the information which directs the functions of a living thing. In that context, the term genetic sequence is often used. Sequences can be read from the biological raw material through DNA sequencing methods.

Nucleic acids also have a secondary structure and tertiary structure. Primary structure is sometimes mistakenly referred to as primary sequence. Conversely, there is no parallel concept of secondary or tertiary sequence.

A Single Nucleotide Polymorphism, also known as Simple Nucleotide Polymorphism, (SNP, pronounced snip; plural snips) is a DNA sequence variation occurring commonly within a population (e.g. 1%) in which a single nucleotide — A, T, C or G — in the genome (or other shared sequence) differs between members of a biological species or paired chromosomes.

Epigenetics is the study of heritable changes in gene function that do not involve changes in the DNA sequence.